A Case report: Abernethy syndrome type-2.

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Abstract:

We are reporting a case of Abernethy syndrome type 2, a rare disorder of the splanchnic venous system. Abernethy syndrome consists of congenital porto-systemic shunts that are a result of persistence of embryonic vessels. A 6 year old child presented to us with generalised weakness involving all four limbs with bilateral ptosis followed by altered sensorium and flapping tremors. Investigations were suggestive of hyperammonemia and coagulopathy. CT angiography of abdomen was suggestive of Abernethy syndrome type 2.

Keywords: Abernethy syndrome, Hyperammonemia, Splanchnic venous system.

Introduction:

Abernethy syndrome is defined as congenital diversion of portal blood away from liver by either end to side or side to side shunt that are a result of persistence of embryonic vessels.[1] Patients have associated anomalies like congenital heart disease, polysplenia, biliary atresia, malrotation, annular pancreas, situs invertus, anomalies of renal tract and skeletal anomalies.[2]

Case History:

A 6 year old male child presented with complaints of altered sensorium, flapping sensorium, weakness of all four limbs, ptosis and lethargy since 10 days and sudden loss of consciousness since 5 hours. Patient was relatively asymptomatic before 10 days. He had lethargy since 10 days and vomiting since 4 days. Then he developed sudden unconsciousness which lasted for 5 hours after which the patient was brought to civil hospital. For the above symptoms patient was prescribed psychiatric medications from a private practitioner. Patient had a past history of hospitalization for cerebral malaria 2 years back. The patient was 2nd of twin, delivered by vaginal delivery with birth weight of 1.5 kg. He was subsequently admitted in NICU for 10 days to ensure proper weight gain.

On examination the patient had altered sensorium, clubbing, flapping tremor of hands and mild pallor. CNS examination revealed hypotonia of all four limbs and plantar reflexes were extensor. Deep tendon reflexes were present. Glassgow coma scale was 12/15. There was no hepatosplenomegaly.

On investigation Hb-9mg/dl, Total count – 11600/cmm, platlet-1,66,000/cmm. Renal function tests were normal. Liver function tests showed a decrease in total protein (4.03 gm/dl) and albumin (2.19 gm/dl). Prothrombin time (22.2 sec)

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and INR (1.91) both were prolonged. Therefore chronic liver disease was suspected.

USG abdomen was done and was suggestive of a vascular channel by which portal vein connected with infrahepatic vena cava. This was suggestive of end to side communication between inferior vena cava and hypoplastic portal vein. MRI Brain was done which was suggestive of hepatocerebral degeneration suggestive of underlying metabolic etiology. Serum ammonia levels were elevated 265.7 micromol/lit (normal range 21-50). The findings were suggestive of Abernethy syndrome type 2. CT angiography of abdomen was done to confirm the diagnosis.

Protein intake was restricted to 1-2 gm/kg/day. Supplements in form of vitamin A, D, E, K and syrup lactulose was given. Fresh frozen plasma was given. Propranolol was started for portal hypertension. Patients sensorium improved, ptosis resolved and prothrombin time normalised with medical management. Gastrosurgical opinion was taken and further management has been planned in conjunction with interventional radiologist and cardiologist in form of closure of abnormal channel by stenting.

Discussion:

Abernethy syndrome type 1a has a separate drainage of superior mesenteric and splenic veins into inferior vena cava, iliac veins or renal veins. Type 1 b has superior mesenteric and splenic veins joining to form a short extrahepatic portal vein which drains into the IVC.[3]

Abernethy type 2 syndrome is a rare disorder of splanchnic venous system characterised by presence of porto-systemic shunts As seen in our patient, Patients with Abernethy type 2 syndrome usually present with digital clubbing, varying degree of dyspnoea on exertion, hepatic encephalopathy and hypogylcemia.[4] Ultrasonography abdomen and CT angiography of abdomen was done which was suggestive of end to side anastomosis between infrahepatic IVC and portal vein.

Management of this includes restriction of protein intake to 1-2 gm/kg/day, vitamin supplements, sodium benzoate for hepatic encephalopathy. Shunt occlusion can be performed either by percutaneous transcatheter coil placement or surgically.

Conclusion:

Although a rare entity Abernethy syndrome can be thought of in patients having hyperammonemia and hepatocerebral degeneration. It can be diagnosed by Ultrasonography of abdomen and confirmed by CT angiography of abdomen.

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